

TOURETTE SYNDROME

What is Tourette Syndrome?

Tourette syndrome (TS) is an inherited, neurological disorder characterized by repeated involuntary movements and uncontrollable vocal (phonic) sounds called tics. In a few cases, such tics can include inappropriate words and phrases.

The disorder is named for Dr. Georges Gilles de la Tourette, the pioneering French neurologist who first described an 86-year-old French noblewoman with the condition in 1885.

The symptoms of TS generally appear before the individual is 18 years old. TS can affect people of all ethnic groups; males are affected 3 to 4 times more often than females. It is estimated that 100,000 Americans have full-blown TS, and that perhaps as many as 1 in 200 show a partial expression of the disorder, such as chronic multiple tics or transient childhood tics.

The natural course of TS varies from patient to patient. Although TS symptoms range from very mild to quite severe, the majority of cases fall in the mild category.

What are the symptoms?

The varied symptoms of TS can be divided into motor, vocal, and behavioral manifestations. Simple motor tics are fast, darting, meaningless muscular events. They can be embarrassing or even painful (such as jaw snapping). They are easily distinguished from simple muscular twitches or rapid fasciculations, e.g., of the eyelid or lip. Complex motor tics may be slower or more purposeful in appearance and more easily described by terms used for deliberate actions.

Complex motor tics can be virtually any type of movement that the body can produce including gyrating, hopping, clapping, tensing arm or neck muscles, touching people or things and obscene gesturing.

At some point in the continuum of complex motor tics, the term "compulsion" seems appropriate for capturing the organized, ritualistic character of the actions. The need to do and then redo or undo the same action a certain number of times (e.g., to stretch out an arm ten times before writing, to "even up," or to stand up and push a chair into "just the right position") is compulsive in quality and accompanied by considerable internal discomfort. Complex motor tics may greatly impair school work, e.g., when a child must stab at a workbook with a pencil or must go over the same letter so many times that the paper is worn thin. Self-destructive behaviors, such as head banging, eye poking and lip biting also may occur. The distinction between complex tics and compulsions may be a difficult one for the physician to make and some "complex tics" may be alleviated by medications used for obsessive-compulsive disorder.

Vocal tics extend over a similar spectrum of complexity and disruption as do motor tics. With simple vocal tics, patients emit linguistically meaningless sounds or noises, such as hissing, coughing or barking. Complex vocal tics involve linguistically meaningful words, phrases or sentences, e.g., "wow," "Oh boy, now you've said it," "Yup, that's it," "but, but..." Vocal symptoms may interfere with the smooth flow of speech and resemble a stammer, stutter or other speech irregularity. Often, but not always, vocal symptoms occur at points of linguistic transition, such as at the beginning of a sentence where there may be speech blocking at the initiation of speech or at phrase transitions. Patients suddenly may alter speech volume, slur a phrase, emphasize a word or assume an accent.

The most socially distressing complex vocal symptom is coprolalia, the explosive utterance of foul or "dirty" words or more elaborate sexual, aggressive or insulting statements (e.g., racial slurs). Coprolalia is not simply obscene speech spoken in anger or to offend. Rather it is often sudden speech (typically just the first syllable of an inappropriate word) that interrupts an otherwise appropriate flow of words. While coprolalia occurs in only a minority of patients with TS (from 5-30%, depending on the clinical series), it remains the most well known TS symptom. A diagnosis of TS does not require that coprolalia be present and the majority of patients do not ever exhibit this symptom.

Some patients with TS may have a tendency to imitate what they have just seen (echopraxia), heard (echolalia), or said (palilalia). For example, the patient may feel an impulse to imitate another's body movements, to speak with an odd inflection or to accent a syllable in just the same manner as another person. Such modeling or repetition may lead to the onset of new specific symptoms that will wax and wane in the same way as other TS symptoms. Some patients also describe "triggers" that almost invariably prompt a tic, e.g., another person coughing in a certain way.

The symptoms of TS can be characterized as mild, moderate or severe by their frequency, their complexity and the degree to which they cause impairment or disruption of the patient's ongoing activities and daily life. For example, extremely frequent tics that occur 20-30 times a minute, such as blinking, nodding or arm flexion, may be less disruptive than an infrequent tic that occurs several times an hour, such as loud barking, coprolalic utterance or touching tics. The premonitory sensory urges tend to be present by 9 to 10 years of age. They are most commonly reported in the shoulder girdle, hands, throat and abdomen.

There may be tremendous variability over short and long periods of time in symptomatology, frequency and severity. Tics typically occur in "bouts" with many tics over a short interval of time. Patients may be able to inhibit or not feel a great need to emit their symptoms while at school or work. When they arrive home, however, the tics may erupt with violence and remain at a distressing level throughout the remainder of the day.

It is not unusual for patients to "lose" their tics as they enter the doctor's office. Parents may plead with a child to "show the doctor what you do at home," only to be told that the youngster "just doesn't feel like doing them" or "can't do them" on command. Adults will say "I only wish you could see me outside your office," and family members will heartily agree.

Often a patient with minimal symptoms may display more severe tics when the examination is over. Thus, for example, the doctor may often see a nearly symptom-free patient who then leaves the office and begins to hop, flail, or bark as soon as he or she reaches the street.

In addition to the moment-to-moment or short-term changes in symptom intensity, many patients have oscillations in severity over the course of weeks and months. The waxing and waning of severity may be triggered by changes in the patient's life; for example, around holidays, children may develop exacerbations that take weeks to subside. Other patients report that their symptoms show seasonal fluctuation. However, there are no rigorous data on whether life events, stresses or seasons do, in fact, influence the onset or offset of a period of exacerbation. Once a patient enters a phase of waxing symptomatology, a process seems to be triggered that will run its course for weeks or months.

In its most severe forms, patients may have uncountable motor and vocal tics during all their waking hours with paroxysms of full body movements, shouting or self-mutilation. At times the tics seem organized in orchestrated patterns that are characteristic of that individual. Despite this, many patients with severe tics manage to achieve adequate social adjustment in adult life, although usually with considerable emotional pain. More than the severity of motor and vocal tics, the factors that appear to be of importance with regard to social adaptation include the seriousness of attentional problems, obsessive-compulsive symptoms, the degree of family acceptance and support, intelligence and ego strength.

In adolescence and early adulthood, patients with TS frequently come to feel that their social isolation, vocational or academic failure and embarrassing symptoms are more than they can bear. At times, a small number may consider and attempt suicide. Conversely, some patients with the most bizarre and disruptive symptomatology may achieve excellent social, academic and vocational adjustment. Fortunately, in many cases, tics diminish during the course of adolescence. However, in other cases (<10%), the tic symptoms can become even more severe in adulthood.

How are tics classified?

There are two categories of tics: simple and complex. Simple tics are sudden, brief movements that involve a limited number of muscle groups. They occur in a single or isolated fashion and are often repetitive. Some of the more common examples of simple tics include eye blinking, shoulder shrugging, facial grimacing, head jerking, yelping, and sniffing. Complex tics are distinct, coordinated patterns of successive movements involving several muscle groups. Complex tics might include jumping, smelling objects, touching the nose, touching other people, coprolalia, echolalia, or self-harming behaviors.

Can people with TS control their tics?

People with TS can sometimes suppress their tics for a short time, but the effort is similar to that of holding back a sneeze. Eventually tension mounts to the point where the tic escapes. Tics worsen in stressful situations; however they improve when the person is relaxed or absorbed in an activity. In most cases tics decrease markedly during sleep.

What causes TS?

Although the basic cause of TS is unknown, current research suggests that there is an abnormality in the gene(s) affecting the brain's metabolism of neurotransmitters such as dopamine, serotonin, and norepinephrine. Neurotransmitters are chemicals in the brain that carry signals from one nerve cell to another.

What disorders are associated with TS?

Not all people with TS have disorders other than tics. However, it is estimated that 50% of the people with TS also have ADHD, where the person has difficulty concentrating and is easily distracted. Other disorders that people with TS may experience include obsessive compulsive behavior, where the person feels that something must be done repeatedly, such as hand washing or checking that a door is locked; learning disabilities, which include reading, writing, arithmetic, and perceptual difficulties; problems with impulse control, which can result in overly aggressive behaviors or socially inappropriate acts; or sleep disorders, which include frequent awakenings or talking in one's sleep.

The wide range of behavioral symptoms that can accompany tics may, in fact, be more disabling than the tics themselves. Patients, families, and physicians need to determine which set of symptoms is most disabling so that appropriate medications and therapies can be selected.

How is TS diagnosed?

Generally, TS is diagnosed by observing the symptoms and evaluating family history. For a diagnosis of TS to be made, both motor and phonic tics must be present for at least 1 year. Neuroimaging studies, such as magnetic resonance imaging (MRI), computerized tomography (CT), and electroencephalogram (EEG) scans, or certain blood tests may be used to rule out other conditions that might be confused with TS. However, TS is a clinical diagnosis. There are no blood tests or other laboratory tests that definitively diagnose the disorder.

Studies show that correct diagnosis of TS is frequently delayed after the start of symptoms because many physicians may not be familiar with the disorder. The behavioral symptoms and tics are easily misinterpreted, often causing children with TS to be misunderstood at school, at home, and even in the doctor's office. Parents, relatives, and peers who are unfamiliar with the disorder may incorrectly attribute the tics and other symptoms to psychological problems, thereby increasing the social isolation of those with the disorder. And because tics can wax and wane in severity and can also be suppressed, they are often absent during doctor visits, which further complicates making a diagnosis.

In many cases, parents, relatives, friends, or even the patients themselves become aware of the disorder based on information they have heard or read in the popular media.

How is TS treated?

Because symptoms do not impair most patients and development usually proceeds normally, the majority of people with TS require no medication. However, medications are available to help when symptoms interfere with functioning. Unfortunately, there is no one medication that is helpful to all persons with TS, nor does any medication completely eliminate symptoms; in addition, all medications have side effects. Instead, the available TS medications are only able to help reduce specific symptoms.

Some patients who require medication to reduce the frequency and intensity of the tic symptoms may be treated with neuroleptic drugs such as haloperidol and pimozide. These medications are usually given in very small doses that are increased slowly until the best possible balance between symptoms and side effects is achieved.

Recently scientists have discovered that long-term use of neuroleptic drugs may cause an involuntary movement disorder called tardive dyskinesia. However, this condition usually disappears when medication is discontinued. Short-term side effects of haloperidol and pimozide include muscular rigidity, drooling, tremor, lack of facial expression, slow movement, and restlessness. These side effects can be reduced by drugs commonly used to treat Parkinson's disease. Other side effects such as fatigue, depression, anxiety, weight gain, and difficulties in thinking clearly may be more troublesome.

Clonidine, an antihypertensive drug, is also used in the treatment of tics. Studies show that it is more effective in reducing motor tics than reducing vocal tics. Fatigue, dry mouth, irritability, dizziness, headache, and insomnia are common side effects associated with clonidine use. Fluphenazine and clonazepam may also be prescribed to help control tic symptoms.

Medications are also available to treat some of the associated behavioral disorders. Stimulants such as methylphenidate, pemoline, and dextroamphetamine, usually prescribed for attention deficit disorders, although somewhat effective, have also been reported to increase tics; therefore their use is controversial. For obsessive compulsive behaviors that significantly disrupt daily functioning, fluoxetine, clomipramine, sertraline, and paroxetine may be prescribed.

Other types of therapy may also be helpful. Although psychological problems do not cause TS, psychotherapy may help the person better cope with the disorder and deal with the secondary social and emotional problems that sometimes occur. Psychotherapy does not help suppress the patient's tics.

Relaxation techniques and biofeedback may be useful in alleviating stress which can lead to an increase in tic symptoms.

Is TS inherited?

Evidence from genetic studies suggests that TS is inherited in a dominant mode and the gene(s) involved can cause a variable range of symptoms in different family members. A person with TS has about a 50-50 chance of passing on the gene(s) to one of his or her offspring. However, that genetic predisposition may not necessarily result in full-blown TS; instead, it may express itself as a milder tic disorder or as obsessive compulsive behaviors or possibly attention deficit disorder with few or no tics at all. It is also possible that the gene-carrying offspring will not develop any TS symptoms. A higher than normal incidence of milder tic disorders and obsessive compulsive behaviors has been found in families of individuals with TS.

Gender also plays an important role in TS gene expression. If the gene-carrying offspring of a TS patient is male, then the risk of developing symptoms is 3 to 4 times higher. However, most people who inherit the gene(s) will not develop symptoms severe enough to warrant medical attention. In some cases of TS, inheritance cannot be determined. These cases are called sporadic and their cause is unknown.

What is the prognosis?

There is no cure for TS; however, the condition in many individuals improves as they mature. Individuals with TS can expect to live a normal life span. Although the disorder is generally lifelong and chronic, it is not a degenerative condition. TS does not impair intelligence. Tics tend to decrease with age, enabling some patients to discontinue using medication. In a few cases, complete remission occurs after adolescence. Although tic symptoms tend to decrease with age, it is possible that neuropsychiatric disorders such as depression, panic attacks, mood swings, and antisocial behaviors may increase.

What is the best educational setting for children with TS?

Although students with TS often function well in the regular classroom, it is estimated that many may have some kind of learning disability. When attention deficit disorder, obsessive compulsive disorder, and frequent tics greatly interfere with academic performance or social adjustment, students should be placed in an educational setting that meets their individual needs. These students may require tutoring, smaller or special classes, and in some cases special schools.

All students with TS need a tolerant and compassionate setting that both encourages them to work to their full potential and is flexible enough to accommodate their special needs. This setting may include a private study area, exams outside the regular classroom, or even oral exams when the child's symptoms interfere with his or her ability to write. Untimed testing reduces stress for students with TS.

Q&A

1. Q. What is Tourette Syndrome?

A. Tourette Syndrome (TS) is a neurological disorder characterized by tics -- involuntary, rapid, sudden movements or vocalizations that occur repeatedly in the same way. The symptoms include:

Both multiple motor and one or more vocal tics present at some time during the illness although not necessarily simultaneously;

The occurrence of tics many times a day (usually in bouts) nearly every day or intermittently throughout a span of more than one year; and

Periodic changes in the number, frequency, type and location of the tics, and waxing and waning of their severity. Symptoms can sometimes disappear for weeks or months at a time.

Onset before the age of 18.

The term, "involuntary," used to describe TS tics is sometimes confusing since it is known that most people with TS do have some control over their symptoms. What is not recognized is that the control, which can be exercised anywhere from seconds to hours at a time, may merely postpone more severe outbursts of symptoms. Tics are experienced as irresistible and (as with the urge to sneeze) eventually must be expressed. People with TS often seek a secluded spot to release their symptoms after delaying them in school or at work. Typically, tics increase as a result of tension or stress, and decrease with relaxation or when focusing on an absorbing task.

2. Q. How would a typical case of TS be described?

A. The term typical cannot be applied to TS. The expression of symptoms covers a spectrum from very mild to quite severe. However, the majority of cases can be categorized as mild.

3. Q. Is obscene language (coprolalia) a typical symptom of TS?

A. Definitely not. The fact is that cursing, uttering obscenities, and ethnic slurs are manifested by fewer than 15% of people with TS. Too often, however, the media seize upon this symptom for its sensational effect.

4. Q. What causes the symptoms?

A. The cause has not been established, although current research presents considerable evidence that the disorder stems from the abnormal metabolism of at least one brain

chemical (neurotransmitter) called dopamine. Undoubtedly, other neurotransmitters, e.g. serotonin, are involved as well.

5. Q. How is TS diagnosed?

A. A diagnosis is made by observing symptoms and by evaluating the history of their onset. No blood analysis or other type of neurological testing exists to diagnose TS. However, some physicians may wish to order an EEG, MRI, CAT scan, or certain blood tests to rule out other ailments that might be confused with TS. Rating scales are available for assessment of tic severity.

6. Q. What are the first symptoms?

A. The most common first symptom is a facial tic such as rapidly blinking eyes or twitches of the mouth. However, involuntary sounds such as throat clearing and sniffing, or tics of the limbs may be initial signs. For a minority, the disorder begins abruptly with multiple symptoms of movements and sounds.

7. Q. How are tics classified?

A. Two categories of tics and several other examples are:

Simple:

Motor -- Eye blinking, head jerking, shoulder shrugging and facial grimacing.

Vocal -- Throat clearing, yelping and other noises, sniffing and tongue clicking.

Complex:

Motor -- Jumping, touching other people or things, smelling, twirling about, and only rarely, self-injurious actions including hitting or biting oneself.

Vocal -- Uttering words or phrases out of context and coprolalia (vocalizing socially unacceptable words).

The range of tics or tic-like symptoms that can be seen in TS is very broad. The complexity of some symptoms is often perplexing to family members, friends, teachers and employers who may find it hard to believe that the actions or vocal utterances are involuntary.

8. Q. How is TS treated?

A. The majority of people with TS are not significantly disabled by their tics or behavioral symptoms, and therefore do not require medication. However, there are medications available to help control the symptoms when they interfere with functioning. The drugs include haloperidol (Haldol), clonidine (Catapres), pimozide (Orap), fluphenazine (Prolixin, Permitil), and clonazepam (Klonopin).

Stimulants such as Ritalin, Cylert, and Dexedrine that are prescribed for ADHD may increase tics. Their use is controversial. For obsessive compulsive traits that interfere significantly with daily functioning, fluoxetine (Prozac), clomipramine (Anafranil), sertraline (Zoloft), risperidone (Risperdal), and paroxetine (Paxil) are prescribed.

Dosages which achieve maximum control of symptoms vary for each patient and must be gauged carefully by a doctor. The medicine is administered in small doses with gradual increases to the point where there is maximum alleviation of symptoms with minimal side effects. Some of the undesirable reactions to medications are weight gain, muscular rigidity, fatigue, motor restlessness and social withdrawal, most of which can be reduced with specific medications. Side effects such as depression and cognitive impairment can be alleviated with dosage reduction or a change of medication.

Other types of therapy may also be helpful. Psychotherapy can assist a person with TS and help his/her family cope, and some behavior therapies can teach the substitution of one tic for another that is more acceptable. The use of relaxation techniques and/or biofeedback can serve to alleviate stress reactions that cause tics to increase.

9. Q. Is it important to treat Tourette Syndrome early?

A. Yes, especially in those instances when the symptoms are viewed by some people as bizarre, disruptive and frightening. Sometimes TS symptoms provoke ridicule and rejection by peers, neighbors, teachers and even casual observers. Parents may be overwhelmed by the strangeness of their child's behavior. The child may be threatened, excluded from activities and prevented from enjoying normal interpersonal relationships. These difficulties may become greater during adolescence -- an especially trying period for young people and even more so for a person coping with a neurological problem. To avoid psychological harm, early diagnosis and treatment are crucial. Moreover, in more serious cases, it is possible to control many of the symptoms with medication.

10. Q. Do all people with TS have associated behaviors in addition to tics?

A. No, but many do have one or more additional problems which may include:

Obsessions which consist of repetitive unwanted or bothersome thoughts.

Compulsions and Ritualistic Behaviors which occur when a person feels that something must be done over and over and/or in a certain way. Examples include touching an object with one hand after touching it with the other hand to "even things up" or repeatedly checking to see that the flame on the stove is turned off. Children sometimes beg their parents to repeat a sentence many times until it "sounds right."

Attention Deficit Disorder with or without Hyperactivity

(ADD or ADHD) occurs in many people with TS. Children may show signs of hyperactivity before TS symptoms appear. Indications of ADHD may include: difficulty with concentration; failing to finish what is started; not listening; being easily distracted; often acting before thinking; shifting constantly from one activity to another; needing a great deal of supervision; and general fidgeting. Adults too may exhibit signs of ADHD such as overly impulsive behavior and concentration difficulties and the need to move constantly. ADD without hyperactivity includes all of the above symptoms except for the high level of activity. As children with ADHD mature, the need to move is more likely to be expressed by restless, fidgety behavior. Difficulties with concentration and poor impulse control persist.

Learning Disabilities may include reading and writing

difficulties, problems with mathematics, and perceptual problems.

Difficulties with impulse control which may result, in rare instances, in overly aggressive behaviors or socially inappropriate acts. Also, defiant and angry behaviors can occur.

Sleep Disorders are fairly common among people with TS. These include frequent awakenings or walking or talking in one's sleep.

11. Q. Do students with TS have special educational needs?

A. While school children with TS as a group have the same IQ range as the population at large, many have special educational needs. Data show that many may have some kind of learning problem. That condition, combined with attention deficits and the difficulty coping with frequent tics, often call for special educational assistance. The use of tape recorders, typewriters, or computers for reading and writing problems, untimed exams (in a private room if vocal tics are a problem), and permission to leave the classroom when tics become overwhelming are often helpful. Some children need extra help such as access to tutoring in a resource room.

When difficulties in school cannot be resolved, an educational evaluation may be indicated. A resulting identification as "other health impaired" under federal law will entitle the student to an Individual Education Plan (IEP) which addresses specific educational problems in school. Such an approach can significantly reduce the learning difficulties that prevent the young person from performing at his/her potential. The child who cannot be adequately educated in a public school with special services geared to his/her individual needs may be best served by enrollment in a special school.

12. Q. Is TS inherited?

A. Genetic studies indicate that TS is inherited as a dominant gene (or genes) causing different symptoms in different family members. A person with TS has about a 50% chance of passing the gene to one of his/her children with each separate pregnancy. However, that genetic predisposition may express itself as TS, as a milder tic disorder or as obsessive compulsive symptoms with no tics at all. It is known that a higher than normal incidence of milder tic disorders and obsessive compulsive behaviors occur in the families of TS patients.

The sex of the offspring also influences the expression of the gene. The chance that the gene-carrying child of a person with TS will have symptoms is at least three to four times higher for a son than for a daughter. Yet only about 10% of the children who inherit the gene will have symptoms severe enough to ever require medical attention. In some cases TS may not be inherited, and cases such as these are identified as sporadic TS. The cause in these instances is unknown.

13. Q. Is there a cure?

A. Not yet.

14. Q. Is there ever a remission?

A. Many people experience marked improvement in their late teens or early twenties. Most people with TS get

better, not worse, as they mature, and those diagnosed with TS have a normal life span. As many as 1/3 of patients experience remission of tic symptoms in adulthood.

15. Q. How many people in the U.S. have TS?

A. Since many people with TS have yet to be diagnosed, there are no absolute figures. The official estimate by the National Institutes of Health is that 100,000 Americans have full-blown TS. Some genetic studies suggest that the figure may be as high as one in two hundred if those with chronic multiple tics and/or transient childhood tics are included in the count.

16. Q. What is the history of TS?

A. In 1825 the first case of TS was reported in medical literature with a description of the Marquise de Dampierre, a noblewoman whose symptoms included involuntary tics of many parts of her body and various vocalizations including coprolalia and echolalia. Later, Dr. Georges Gilles de la Tourette, the French neurologist for whom the disorder is named, first described nine cases in 1885. Samuel Johnson, the lexicographer, and Andre Malraux, the French author, are among the famous people who are thought to have had TS.

17. Q. What is the current focus of research?

A. Since 1984, the TSA has directly funded important research investigations in a number of scientific areas relevant to TS. Recently, studies have intensified to understand how the disorder is transmitted from one generation to the next, and researchers are working toward locating the gene marker for TS. That focus has been enhanced by the efforts of a TSA- supported international group of scientists who have formed a unique network to share what they know about the genetics of TS and to systematically cooperate to unravel the unknown.

Additional insights are being obtained from studies of large families (kindreds) with numerous members who have TS. At the same time, investigators continue to study specific groups of brain chemicals to better understand the syndrome and to identify new and improved medications.